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Kallmann's Syndrome and Transsexualism

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Until the present, in the world literature only one patient with Kallmann's syndrome has been reported who became transsexual. This patient was seen almost 50 years ago. In this report, a second case is presented to encourage studies of the sexual and gender identity development in these patients. This patient's rare endocrine disorder and secondary emotional problems have led to negative consequences because appropriate treatment of her transsexualism became impossible.

KEY WORDS: Kallmann's syndrome; hypogonadism; gender identity disorder; transsexualism.

INTRODUCTION

Kallmann's syndrome is an inherited disorder characterized by hypogonadotropic hypogonadism and anosmia (Kallmann *et al*., 1944). Hypogonadism is due to deficiency of gonadotropin-releasing hormone and anosmia is due to hypoplasia or aplasia of the olfactory bulbs and tracts. In Western countries, a prevalence of 1 in 10,000 to 1 in 60,000 persons is reported with an excess of male over female patients suggesting X-linked inheritance. Genetic studies have localized X-linked Kallmann's syndrome at Xp 22.3 (Duke *et al*., 1995; Hardelin and Petit, 1995; Meitinger *et al*., 1990).

Typically, male patients show eunuchoid growth after delayed or absent puberty, infantile genitals, sparse body hair growth, high-pitched voice, and hyposmia or anosmia associated with nasal saddle deformities. A large number of neurologic deficits and other somatic defects have been described (e.g., Bick *et al*., 1992), but

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the literature is void of describing psychosexual and gender identity development in these patients although problems in this area are likely to be present.

Since the publication of Kallmann *et al*. in 1944 only one case of Kallmann's syndrome with atypical gender identity development has been described in the literature. Bürger-Prinz *et al*. (1966) reported on a 23-year-old male patient who presented in the early 1950s with a wish for sex reassignment surgery. The patient reported that he had felt like a girl since early childhood and preferred doll play and female activities. At the age of 15, he engaged sexually with older boys. Later on, when puberty did not occur, he began cross-dressing. Sexual activities were reduced to masturbating once or twice a month. Heterosexual interests were absent. Laboratory tests showed markedly decreased gonadotropic hormones. Testicular biopsy showed atrophic tubuli. Leydig cells were absent; bilateral anosmia was present.

The patient was treated with gonadotropic hormones. His physical appearence rapidly virilized. It is not explained how administration of gonadotropic hormones could lead to virilization, given the biopsy results. It appears likely that Leydig cells were only diminished in number, but were not absent. After 2 months, the patient's transsexual wishes subsided; heterosexual interests appeared; and masturbatory activity increased. He then engaged in a sexual relationship with an older woman. A 17-year follow-up (Fischer, 1971) showed a persistent absence of transsexual symptoms; the patient lived for 10 years in a stable sexual relationship with a woman.

CASE REPORT

We report a 58-year-old genetically male patient, L. B., who presented 20 years ago with a wish for sex reassignment surgery. Since the age of 39, L. B. has lived continuously as a female. (For reasons of clarity we refer to the patient throughout her whole course of life as "she.") Medical history and examination showed Kallmann's syndrome with some atypical features. Since the age of 38, the patient was frequently misdiagnosed (e.g., paranoid schizophrenia) and, in consequence, had extreme hardships like involuntary psychiatric hospitalization. Sex reassignment surgery has not been possible so the patient continues to live "in limbo" (L. B.). She was seen first by one of the authors (B. M.) when her 7-year-old daughter T. was referred for psychotherapy to help her deal with her father's sex change. T. was dealing well with this problem, but severe psychiatric problems became soon apparent in L. B. (e.g., depression and personality disorder). Psychotherapy was started for 1 year until her therapist moved abroad. Concomitantly, the patient's wife and daughter were seen on a regular basis. Until now, yearly follow-up studies were done with L. B. for 16 years.

The patient is a 168-cm tall white genetic male, weighing 142 lbs, with feminine habitus. She has a high-pitched voice, which sounds hoarse due to heavy

smoking. Her skin is fine, there are fine wrinkles around eyes and mouth, a normal female hair-escutcheon. Axillary hair was shaved, pubic hair is female in pattern. There is no male muscle development. Her chest shows scars following four gynecomastia operations at the age of 18–30. There is a scar over the right hip; bone chips were removed to reconstruct nasal saddle deformity. Bilateral anosmia is present. Aureola and nipples are female in structure and pigmentation. The penis measures about 5 cm and has a normal urethral opening. A perineal aperture was not found. Testicles were tender and firm, palpable bilaterally in the inguinal canals measuring 1–2 cm in diameter and 4 ml in volume. The scrotum appeared rugated and is not bifid.

The patient reported recurrent painless bloody urethral discharge occuring since the age of 27 every 4–5 weeks, lasting 24 hours, an unexplained phenomenon reported rarely also by other transsexual patients. Genetic studies showed a male karyotype (46, XY). Endocrine studies at the age of 38 showed low, castrate-levels of testosterone without elevated gonadotropins and a rise of testosterone from 0.07 to 0.96 ng/ml after HCG-stimulation. There are reports of very high estradiol levels (320, 1920, 1480, and 3570 pg/ml), the source of which remains unknown. L. B. has persistantly denied estrogen intake. Endocrine studies were repeated at the age of 42. Estradiol levels were low, below 0.1 pg/ml, rising to 18.0 pg/ml after HCG-stimulation (5000 U HCG qd $\times$ 5 d). Testosterone levels remained low ($<$0.1 ng/ml) even after HCG-stimulation. LH-FSH-levels were low (LH 5.8 mU/ml, FSH 13.4 mU/ml), rising to normal levels after HCG-stimulation (LH 88.1 mU/ml, FSH 58.9 mU/ml). Prolactin levels were high (16.8 mg/ml).

CT scan of abdomen and pelvis showed no abnormal masses in the region of the adrenal gland; no uterine tissue was identified in the pelvis. A prostate gland of normal size was seen in normal position with focal areas of low attenuation. In the region of the inguinal canals, two oval soft tissue masses were seen consistent with bilaterally undescended testicles. CT scan of sella showed the sella turcica of normal size; no mass lesions were identified within the sella or in the suprasellar cistern.

Past History

Past history data could only be obtained from the patient herself and her wife because relatives could not be reached. However, past history data given during prior hospital admissions and to prior therapists showed high coincidence in details.

L. B. is the third child of a white Caucasian mother and an American Indian father. At the age of 1, she was given to foster parents for 1 year. Then she lived until the age of $13^1/_2$ years in an orphanage. Her two older sisters stayed with her mother after the parents divorced. Her father abused alcohol. Major diseases in the family are unknown to the patient. At the age of 14–17, the patient was sent

to a reform school. After school, the patient served in the Air Force for 2 years and was discharged because of concealing female clothes in a foot locker. L. B. explained to the authorities that she kept these clothes for her girlfriend. Later the patient worked as a musician and ran a radio and TV repair shop. During the year prior to starting psychotherapy she worked as a doorman.

L. B. reports that she has always felt confused about her gender identity. She dates wishes to be female to the age of 4 or 5, when she was caught observing girls in the orphanage where boys and girls lived separately. For punishment she was forced to live dressed as a girl with the other girls for some days. L. B. has always held this as the reason for her gender identity confusion. Later on her physical development began to contribute to the problem. Puberty was delayed. She never developed a male voice. Female breast tissue grew at the age of 17 and was surgically removed four times because L. B. tried desperately to live and pass as male. She started relationships with two women who became pregnant. The oldest child is a boy, presumedly fathered by L. B. at the age of 30; the boy lives with his mother. L. B. states she is not sure about her being the father. Her medical condition (undescended testicles) appears also to be incompatible with fertility. The second child, a girl, was born when the patient was 34. L. B. continues to live with the daughter's mother whom she married while still living in the male role. There are doubts also about L. B. being the father of this child. The patient reports that she has had difficulties in performing sexual intercourse. During intercourse she experienced painful feelings in her penis; only a few drops of watery fluid were emitted. Sperm counts could not be made. Further examination of her undescended testicles were refused.

After trying desparately to live and function as a male until her late thirties, L. B. felt increasingly insecure about her gender identity. At the age of 39, the patient was referred to a major medical center for psychological and endocrine work-up. Repeatedly, L. B. became emotionally upset and suicidal, once setting fire to herself, because she felt treated like a "freak" or "pervert." This led to three subsequent psychiatric admissions where diagnoses of chronic paranoid schizophrenia, adjustment disorder with depression, and borderline personality disorder were made.

L. B.'s endocrine problems were attributed to exogenous intake of estrogens. Sex reassignment surgery was never considered feasable by her prior therapists. Meanwhile, the patient's wife had sought counselling by a child psychiatrist because she was then worried about her 7-year-old daughter's development. Although the daughter was found dealing exceptionally well with this situation, it soon became apparent that her father, L. B., was in urgent need of further psychotherapy.

Course of Therapy

Supportive psychotherapy was started with sessions twice weekly. The review of her past history and past examinations was painful for L. B. Her most urgent

wish was now to become completely female, to have sex reassignment surgery. Because her case was highly unusual, great caution was taken not to make premature decisions. Administration of estrogens was refused by the patient. L. B. became increasingly depressed and suicidal. She spent whole sessions crying and screaming in pain, complaining that nobody was willing to help her and everybody treated her as a freak. Tricyclic antidepressant medication gave her only minor relief. Three months after starting therapy, L. B. again became seriously suicidal so that hospitalization was necessary. During this time, thorough medical studies were done with the results reported here. When confronted with the results of medical and psychiatric evaluations, that is, Kallmann's syndrome and transsexualism (according to DSM-III classification), L. B. very angrily refused the latter diagnosis. She remains convinced until now that her endocrine problems are the reason for her gender identity disorder. Subsequently, the patient was referred to a urologist experienced in sex reassignment surgery. Here L. B. felt being treated like a "common transsexual" and became so upset that she left the office, screaming and sobbing loudly. The urologist refused to see this emotionally unstable patient again; an experience L. B. has repeated with other psychiatrists also.

In her 1-year long therapy, it became evident that she had felt her gender identity disorder to be present since early childhood. Surprisingly, she reported in one more quiet session that she was unaware of the anatomical sex difference until the age of 15 when she asked a girl to show her genitals to her. When the interpretation was given that it was too painful for her to acknowledge the sex difference openly because she always felt like a female but knew very well about her genitals being male, she responded with shock, anger, and irritation. Her memories of the orphanage, where she tried to observe girls secretly, demonstrate that L. B. was driven early to find a solution for her gender identity problem.

During the past 14 years, L. B.'s situation has remained essentially unchanged. Attempts to continue the therapy by a psychiatrist experienced in treating patients with gender identity disorders failed because L. B. continued to demand immediate sex reassignment surgery. With increasing age L. B. has resigned to living "in-between," in her view as a full-time woman with male genitals. She has never left her residential neighbourhood where she is well known. Painful teasing, frequent in the past, has stopped.

At the age of 52, 9 years after terminating psychotherapy, bilateral inguinal orchidectomy was performed. Only fibroid tissue was found, and no further histologic studies were done. At this time, she was put on estrogens by her family doctor. Testosterone and estradiol levels continued to be low before surgery (estradiol <10 ng/l, testosterone <20 ng/ml). Gonadotropin levels were found to be low as well: LH <0.3 U/l, FSH <0.3 U/l, TSH 0.18 MU/l. Meanwhile (2000.2) L. B. has given up the wish for sex reassignment surgery stating it would no longer be of any use. She continues to live with her wife and daughter whose successful career is a source of major pride for her. Her mental state has improved greatly, she is no longer prone to depression and negative emotional outbursts.

DISCUSSION

Cases like this raise the question of the significance of biologic forces in the development of gender identity disorders. Sociogenetic studies emphasize today how much transsexuals have been shaped as a certain species throughout the centuries by medical science, especially by psychiatry and surgery, and how much social recognition they have gained in Western countries in the unfolding of a "neosexual revolution" (Sigusch, 1998), even including special laws in several European countries.

In the case of L. B., the specific etiology of her transsexualism remains open. It is likely that co-occurrence of traumatic childhood events, for example, being separated from parents and being given away into an orphanage, and biologic factors due to her Kallmann's syndrome led to gender identity disorder. DSM-IV diagnostic criteria for gender identity disorder demand absence of a physical intersex condition. The appropriate diagnosis would now be gender identity disorder not otherwise specified, examples include intersex conditions with accompanying gender dysphoria. ICD-10 diagnostic criteria, however, allow diagnosis of transsexualism (F 64.0) because accompanying Kallmann's syndrome does not constitute an exclusion criterion.

The presence of Kallmann's syndrome and also a very unstable mental condition in the case of L. B. led to the result that sex reassignment measures were never seriously considered and later on even refused forcing her to continue to live "in-between," Furthermore, L. B. considered her endocrine disorder as the only reason for her gender identity disorder. She always refused to consider herself as transsexual. As a result she found herself "stuck in the middle." Because of this stressful situation, she became emotionally even more disturbed.

It might be hypothesized that living "in-between" without sex reassignment has enabled L. B. to continue to live with her family; living as a "real" woman might have brought on even more problems for her and her family. Her quiet acceptance of her fate now at the age of 59 appears to support this different point of view that at least L. B. was highly ambivalent about going all the way to become fully female, a phenomenon also seen in therapies with other transsexual patients (Meyenburg, 1999).

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